NEBRASKA NEWBORN SCREENING PROGRAM

Volume 1, Issue 1

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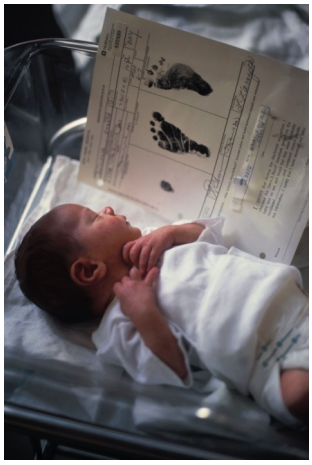
Health Care Provider's Update Newborn (Blood-spot) Screening



Update Contents

Cystic Fibrosis Screening Test Changes

The Nebraska NBS Program, Advisory Committee and PerkinElmer Genetics laboratory, continually evaluate ways to assure the highest quality screening for our babies.

Consistent with our quality improvement practices, the screening test for cystic fibrosis will change effective July 6, 2010. This will decrease (but not eliminate) the likelihood of missing affected newborns.

Previously the Immuno-reactive Trypsinogen (IRT) test would reflex to look for the ΔF508 mutation when the IRT was ≥ 90 ng/mL. If no ΔF508 was found, but the IRT was actually ≥ 130 ng/mL, the laboratory would request a repeat newborn screening specimen.

This will now change so that if no ΔF508 is found, all babies with IRT's ≥ 90 ng/mL will have a repeat specimen requested. If on the repeat specimen, the IRT continues to be ≥ 90 ng/mL, it will then reflex to the x-TAG 39+ mutation panel.

Approximately 2 more babies per week statewide will need to have repeat specimens collected under the new protocol which will help avoid the near-miss situation encountered. In that case the baby had a 125 ng/mL on the initial IRT and no ΔF508. No repeat was requested by the lab or program but thanks to the physician recalling cautionary remarks of Dr. Colombo, the Director of the CF Center, a repeat was ordered and baby was diagnosed and treated early.

Screening Test Changes for Congenital Adrenal Hyperplasia

Test results will look a little different, starting with babies' specimens arriving at the lab on July 6, 2010. There will be new weight-range adjusted cut-offs for the 17-Hydroxyprogesterone (17-OHP). Screen results with elevated 17-OHP will continue to reflex to the extracted 17-OHP but this will also have a different cut-off.

The laboratory has closely evaluated the data over time, and this change is being made to more accurately reflect the performance of the assay, maintain relatively low numbers of false positives, and maintain confidence that we will identify babies affected with salt wasting congenital adrenal hyperplasia. (As before, not all forms of CAH will be detected by this screening assay). Changes are being made with approval of the Newborn Screening Advisory Committee and the Program.

Inside this issue:

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New lab report information for Premature babies for Congenital Primary Hypothyroidism Screening Results

"We need to be able to assure a quality screening test, follow-up and access to treatment, when considering addition of new diseases to the screening panel", James Harper, MD, Pediatric Hematologist, Chair Nebraska Newborn Screening Advisory Committee.

Screening for Congenital Primary Hypothyroidism (CPH) is done by testing the blood spot for Thyroxin (T4) on all babies, followed by TSH on the lowest 10% of T4's in the run at the laboratory.

It has been well documented that premature and low birth weight newborns tend to have lower T4's than term or normal birth weight babies. These babies tend to account for a disproportionately high percent of babies requiring repeat or confirmatory testing for CPH.

In an effort to provide physician's with a better picture of the actual distribution of T4's in newborns of various birth weights, a new table will show on the laboratory results for all babies who had TSH testing done. The table will show the mean average, and lowest tenth percentile for various low-birth-weight ranges where clear distinctions can be made from the data.

This data can be used by the physician to help judge relative risk based on the baby's birth weight and actual results. For example a 1.3 kg baby with a T4 of 10.1 and a TSH of 25.8 on a greater than twenty-four hour specimen will be presumptive positive, and need a confirmatory test of serum Free T4 and TSH. In addition the table will show:

Birthwt	Mean	10%
< 1.26kg	8.6	4.0
1.26-1.5kg	10.2	6.0
1.51-1.75kg	11.7	7.0
1.76-2kg	13.0	9.0
2.01-2.5kg	15.8	10.0

Federal Health and Human Services Secretary Endorses Screening for SCID

May 21, 2010, Washington DC. The Secretary of the Department of Health and Human Services adopted the recommendation of the Advisory Committee on Heritable Diseases in Newborns and Children regarding SCID (Severe Combined Immune Deficiency). This recommendation includes SCID as a "Core Condition" in the Recommended Uniform Screening Panel to be screened, and includes related T-cell lymphocyte deficiencies in the list of secondary targets to be reported.

Responding to this national recommendation, Nebraska's Newborn Screening Program and Advisory Committee have begun evaluating capacity of our system (education/testing/follow-up/ confirmation & diagnosis/ treatment & management services) to be able to add this group of conditions to the screening panel. PerkinElmer Genetics, Inc., Nebraska's contracted newborn screening laboratory has the screening test available.

The Screening Test for SCID

SCID and related T-cell lymphocyte deficiencies have been screened under a pilot/research status in Wisconsin and Massachusetts for nearly two years. The DNA based test (PCR) looks for low numbers of TRECS (T-cell receptor excision circles) as an indicator of low T-cell production. Babies with low numbers of TRECS are referred for further testing.

At the May 2010 National Newborn Screening and Genetics Symposium these programs reported on their experience of screening for SCID. They had identified several babies with various T-cell lymphocyte deficiencies that are responsive to early treatment (usually bone marrow stem cell transplant) and one baby with classical SCID.

Early diagnosis and treatment is essential to the healthy survival of these babies.

The test at PerkinElmer Genetics, Inc. is also a DNA (PCR) based test, that has been validated. The laboratory reports having optimized the test to maximize sensitivity and specificity. Regulatory approval would be needed for mandatory screening in Nebraska. The Advisory Committee will continue to address this.

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Left-Over Dried Blood Spots (DBS), How can they "Really" be Used?

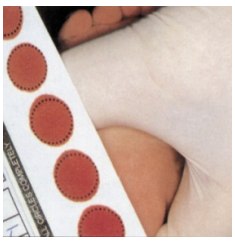
There has been much media coverage about how the left over blood specimens can be used. Lawsuits in Texas and Minnesota have been resolved resulting in destruction of millions of stored blood spots in Texas and changes in laws in both states.

Health care providers can educate new parents about how the blood spots are used from Nebraska births. The "Parent's Guide To Your Baby's Newborn Screening" (provided to all birthing hospitals), has informed parents for many years, that the blood spots are stored for 90 days and then must be destroyed. They can only be used for research with the parent's consent and in compliance with Federal regulations on Institutional Review Board approval.

Nebraska's Advisory Committee strongly supports that use of the residual DBS must be protected primarily for the newborn's benefit during the 90 day storage period. One particular use could be retrieving it to test for congenital cytomegalovirus for babies with confirmed hearing loss to help determine etiology.

There is little disagreement nationally that these DBS are a valuable resource. However, there is yet to be a national consensus on the standards for how they can be used and who has rights to the specimens.

The Secretary's Advisory Committee on Heritable Diseases in Newborns and Children has a work group developing a briefing paper.



"The new guidelines will help standardize how we get screening done efficiently, with the least number of specimens for our NICU babies.". Khalid Awad, MD, Neonatologist, Vice Chair Nebraska NBS Advisory Committee and member of the workgroup that developed the CLSI Guideline.

Newborn Screening Regulation Changes Proposed

The Nebraska Newborn Screening Advisory Committee has recommended changes to the Newborn Screening regulations that will adopt practices consistent with new national guidelines. The new guidelines developed by the Clinical and Laboratory Standards Institute, address "Newborn Screening for Premature, Low Birth-weight and Sick Newborns." The guidelines recommend practices for these babies which account for about 10% of all births, but between 40%-75% of all necessary follow-up activity.

The proposal would continue the requirement that a specimen be collected prior to transferring a baby to a NICU. In addition it would require the NICU to verify that collection has taken place and if not, to obtain a specimen before *any* treatment (except respiratory) has occurred.

If, after the public regulation revision process, the revisions are adopted, educational sessions are planned for all Nebraska NICU's.

Access to Updates on Newborn Screening

In the spirit of stewardship for all of our tax dollars, the Nebraska Newborn Screening Program is asking for your help. We have substantially reduced our printing expenses, by no longer producing a quarterly newsletter, and no more printing of the Annual Report.

However, we need to be able to keep you, the health care provider informed of changes that affect your patients and your practice. Important changes do not usually occur more frequently than annually.

An easy way for automatic notification is to sign up for e-mail notification when updates are posted to our web page. You can select which areas from the Department of Health and Human Services web-page you want to receive updates on. For example, Newborn Screening, Immunizations, and Perinatal and Infant Health might be the only three, or you can select from dozens of other helpful links. To sign up, go to www.dhhs.ne.gov/

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Nebraska Newborn Screening Program

Visit us on the web. We have a wealth of information here about Nebraska's program, requirements and many links to trusted sources.

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Sign up today, for automatic notice of updates to newborn screening information!

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